Recombinant Human WFS1

Catalog No: #GP12544

Package Size: #GP12544-1 100ug



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Description

Product Name	Recombinant Human WFS1
Brief Description	Recombinant Protein
Immunogen Description	Fusion protein corresponding to a region derived from 691-890 amino acids of human WFS1
Target Name	Wolfram syndrome 1 (wolframin)
Other Names	WFS; WFRS; WFSL; CTRCT41
Accession No.	Swissprot:O76024Gene Accession:BC030130
Uniprot	O76024
GenelD	7466;
Storage	-20~-80°C, pH 7.6 PBS

Background

This gene encodes a transmembrane protein, which is located primarily in the endoplasmic reticulum and ubiquitously expressed with highest levels in brain, pancreas, heart, and insulinoma beta-cell lines. Mutations in this gene are associated with Wolfram syndrome, also called DIDMOAD (Diabetes Insipidus, Diabetes Mellitus, Optic Atrophy, and Deafness), an autosomal recessive disorder. The disease affects the brain and central nervous system. Mutations in this gene can also cause autosomal dominant deafness 6 (DFNA6), also known as DFNA14 or DFNA38. Alternatively spliced transcript variants have been found for this gene.

References

Note: For in vitro research use only, not for diagnostic or therapeutic use. This product is not a medical device.

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